Radiology Case Reports 15 (2020) 1604–1608

Available online at www.sciencedirect.com

ScienceDirect

journal homepage: www.elsevier.com/locate/radcr

Case Report

# Tracheal agenesis clinical presentation in a preterm infant: Prenatal MRI difficulties and autopsy findings ☆,☆☆

*Said Saadi, MD*[a,b,*], *Nouha Ben Abdeljelil, MD*[a,c], *Amina Ben Salem, MD*[a,d], *Fatma Zohra Chioukh, MD*[a,e], *Nidhal Haj Salem, MD*[a,b]

[a] *Faculty of Medicine, University of Monastir, Tunisia*
[b] *Department of Forensic Medicine, Fattouma Bourguiba Teaching Hospital, 5000, Monastir, Tunisia*
[c] *Department of Pathology, Fattouma Bourguiba Teaching Hospital, 5000, Monastir, Tunisia*
[d] *Department of Medical Imaging B, Maternity and Neonatal Teaching Center, 5000, Monastir, Tunisia*
[e] *Department of Intensive Care and Neonatal Medicine, Maternity and Neonatal Teaching Center, 5000, Monastir, Tunisia*

ARTICLE INFO

*Article history:*
Received 28 May 2020
Revised 24 June 2020
Accepted 25 June 2020

*Keywords:*
Tracheal agenesis
Congenital abnormalities
Prenatal diagnosis
Magnetic Resonance Imaging
Autopsy

ABSTRACT

We describe, the clinical presentation of a rare case of Tracheal Agenesis in a preterm infant and we highlight magnetic imaging resonance (MRI) and autopsy findings to better characterize this anomaly. A 30-year-old female presented for acute polyhydramnios at 30 weeks gestation of a male foetus. Prenatal MRI was performed and excluded this diagnosis. After delivery, the neonate presented a respiratory distress. The laryngoscopy control of tube position concluded to an esophageal intubation. A second reading of antenatal MRI was made. An autopsy was performed. The internal examination of the organs revealed broncho-oesophageal fistula. The upper airways were obstructed at the larynx. Fetal MRI should be interpreted with caution when Tracheal Agenesis is highly suspected.



☆ Funding: None.
☆☆ Competing Interests: The authors have declared that no competing interests exist.
* Corresponding author.
E-mail addresses: Said.saadi1755@gmail.com (S. Saadi), nouhaba@yahoo.fr (N. Ben Abdeljelil), bensalem_amina@hotmail.fr (A. Ben Salem), zohra_chioukh@yahoo.fr (F.Z. Chioukh), nidhal.hajsalem@yahoo.fr (N. Haj Salem).
https://doi.org/10.1016/j.radcr.2020.06.047

## Introduction

Tracheal Agenesis (TA) is a rare airway congenital anomaly with an incidence of 1 per 50,000 newborns [1]. It is characterized mainly by the partial or total absence of the trachea. It may or may not be associated with a Tracheo-Osophageal Fistula (TOF). Despite the progress made in surgical techniques, the prognosis is poor [2]. In prenatal diagnosis, tracheal agenesis may go unnoticed. Thus, we describe, the clinical presentation of a rare case of tracheal agenesis in a preterm infant and we highlight magnetic resonance imaging (MRI) and autopsy findings to better characterize this anomaly.

## Case report

A 30-year-old female was transferred to a level 3 maternity for acute polyhydramnios at 30 weeks gestation. An obstetrical ultrasound suspected oesophageal atresia. Prenatal MRI was performed and excluded this diagnosis. Two weeks later, the patient was transferred to the delivery room for a retroplacental hematoma. Foetal extraction was immediately decided, and the patient gave birth through caesarean section to a living male fetus, weighing 1360 g. Apgar score was 3 at 1 minute. At birth, the neonate had no cry and persistent apnea despite stimulation.

Despite positive pressure bag mask ventilation, oxygen saturation failed to improve. The baby was intubated and transferred to the neonatal intensive care unit. Initial venous blood gas measurements were pH = 7.00, $PCO_2$ = 67 mm Hg, bicarbonate 16 mmol/L, and lactate 8.1 mmol/L. On day 3 of life, persistent hypercapnia was noticed ($PCO_2$ = 110 mm Hg) despite high peak inspiratory pressure (24 $cmH_2O$). Echocardiography was normal. Chest X-Ray revealed normal pulmonary parenchyma, an intragastric tube, and abdominal distention (Fig. 1). The laryngoscopy control of tube position concluded to an esophageal intubation. The intubation attempts by the neonatologist failed because of a tracheal agenesis. A Fiberoptic bronchoscopy was impossible and he died rapidly.

Given the strong suspicion of this tracheal anomaly, a second reading of antenatal MRI was made and led to the diagnosis of tracheal agenesis with Broncho-oesophageal fistula (BOF; Fig. 2). An autopsy was requested by the attending neonatologist to better support the diagnosis. The internal examination of the organs revealed a BOF 8 cm from the upper extremity of the oesophagus and 5 cm from the oesophageal hiatus. The upper airways were obstructed at the larynx (Fig. 3). The trachea was absent. The rest of the organs were macroscopically normal. The postmortem histological examination confirmed the presence of a fistula between the oesophagus and the left bronchus (Fig. 4). Indeed, there is a communication between a communication between the bronchial and oesophageal lumens. Samples taken from the lungs showed a significant haemorrhage of lung tissue. The samples taken from the rest of the organs were normal.

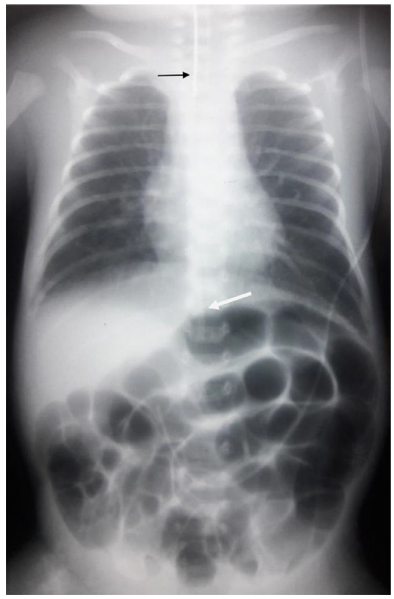

**Fig. 1 – Chest X-ray day of life 1 shows an intra tracheal tube (black arrow), intra gastric tube (White arrow) and abdominal distension.**

## Discussion

TA is an unusual and rare fetal malformation with a prevalence of 1 in 50,000 newborns and with a male predominance. It can be suspected during pregnancy by hydramnios, as in our case. However, the prenatal diagnosis is difficult and only few cases have been reported. Thus, 149 cases were reported [3]. Among the reported cases, mortality was estimated at of 85% [2]. Tracheal agenesis is present mainly in the context of an intrinsic obstruction of the upper airways such as laryngeal agenesis. The thinning of the alveolar ripe, the decrease in the number of pneumocytes type II as well as the decrease in the production of the surfactant are the most described parenchymal lesions in this case [4,5]. The presence of these different anomalies is responsible for a hyper-expansion of the lungs. The radiological abnormalities usually found in MRI are increased signal of the lungs, enlargement and flattening of the diaphragmatic cupolas as well as dilation of the airways upstream of the obstruction level, massive ascites and a deviated heart and compressed with an increase in the volume of the placenta. In our case, these anomalies were not identified.

Postnatal diagnosis is based on the recognition of specific clinical signs in the newborn with TA: respiratory distress with breathing movement without appropriate air entry, no audible cry, and failed endotracheal intubation. As the case illustrates, the intubation in the delivery room was oesophageal and the survival was possible because of BOF. Recently, a life-saving esophageal intubation in a neonate with undiagnosed tracheal agenesis has been reported. This 34-week-old premature infant with imperforate anus required loop ileostomy surgery and TA was diagnosed after a nonairway-related procedure [6].

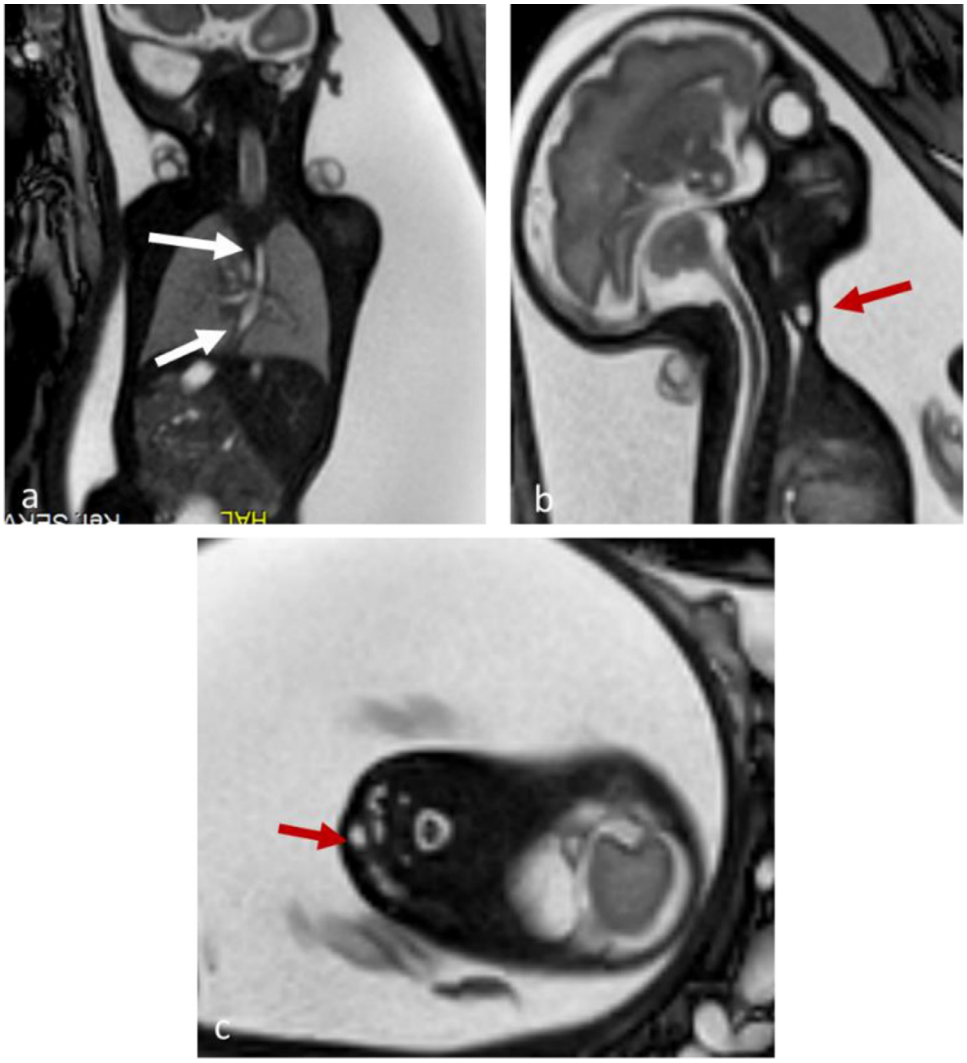


**Fig. 2 – Fetal MRI at 26-weeks gestation. Coronal (a), sagittal (b) and axial (c) T2-weighted images shows a subjectively large amount of amniotic fluid surrounding the fetus. The bronchus arises from the esophagus (white arrow) separately (a). A tiny amount of airway fluid (red arrow b, c) is seen below the epiglottis with no discernible trachea visualized below this level.**

Postmortem examination revealed the presence of communicating unilateral left bronchus and an obstruction cartilage below the larynx. Tracheal tissue was absent. When a BOF is absent, TA can produce signs and symptoms that allow prenatal diagnosis of hyperechoic enlarged Lungs, filled with fluid, dilated trachea and bronchi with absent flow in the trachea during respiration with or without cardiac dysfunction, diaphragmatic flattening, and ascites are found with TA without BOF. This constellation of findings is known as called Congenital Obstruction High Airway Obstruction Syndrome. Fetal MRI can confirm the diagnosis in utero in case of suspicion. The most common abnormality in antenatal TA is polyhydramnios, but it is nonspecific and may be associated with many congenital abnormalities. Because this patient had a tracheoesophageal fistula, abnormalities of the Congenital Airway Obstruction Syndrome were absent. For our foetus, the interpretation of MRI was difficult and was repeated because of the strong suspicion of the diagnosis. The large variation in tracheal malformations, especially in the absence of TOF, led to the development of 2 classifications that are the most commonly used for TA, by Floyd and Faro [7,8]. The Faro classification is the one used for this case because of its greater accuracy. Our case is of type C according to the classification of faro which represents 46.6% of the cases. TA type 2 and 3 generally are still considered unamenable to surgical repair [9].

The possibility of diagnosing TA prenatally allows for an appropriate change in the counselling and management of pregnancy. The prenatal aspect of Congenital Obstruction High Airway Obstruction Syndrome is a less frequent presentation, and technical assistance should be considered when there is polyhydramnios, especially in association with other abnormalities. MRI and medical genetic tests are useful additions to the prenatal check-up.

Several studies have disconcerted the association of TA with other congenital malformations. Smith et al [3] found 94.2% of 194 cases of AT, concomitant malformations associated with TA. The most found malformations are VACTREL and TARCD [10]. In the present case, except for congenital

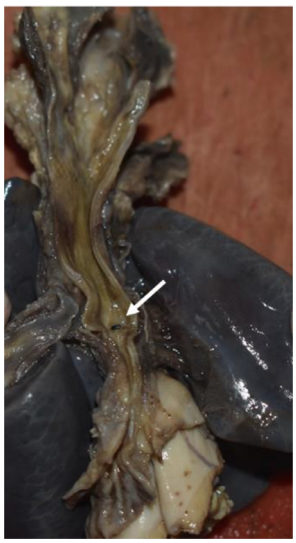

**Fig. 3 – Macroscopic appearance of the fistula between the left lung bronchus and the oesophagus (white arrow).**

laryngeal obstruction, no malformations were found by clinical examination or autopsy.

## Conclusion

Clinicians should be aware of TA which can be prenatally suspected in case of polyhydramnios or other congenital malformations. An MRI with a careful interpretation is important for the confirmation and can help, in this case, to guide therapeutic decisions and/or stop pregnancy on time.

## Ethics approval

All procedures performed in this study (involving human) were in accordance with the ethical standards of the Research Ethics Committee of the Faculty of Medicine, University of Monastir and with the 1964 Helsinki declaration and its later amendments or comparable ethical standards.

## Informed consent

Informed consent was obtained from the parents of the deceased neonate.

## Authors' contribution statements

All authors contributed to the study conception and design. Conceptualization and supervision were performed by Amina BEN SALEM, Fatma Zahra CHIOUKH and Nidhal HAJ SALEM. Material preparation, data collection and analysis were performed by Said SAADI, Nouha BEN ABDELJELIL. The first draft of the manuscript was written by Said SAADI and all authors commented on previous versions of the manuscript. All authors read and approved the final manuscript.

## Acknowledgments

None.

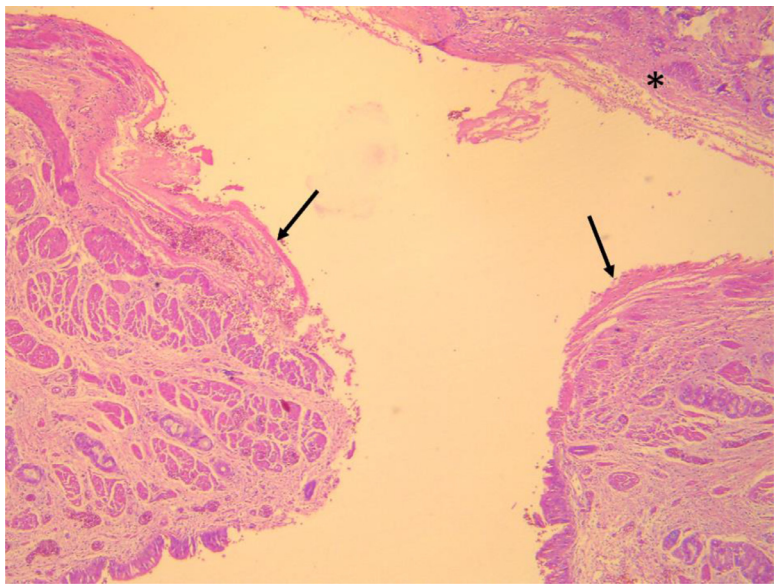


**Fig. 4 – Wide communication between bronchial lumen (*) and the oesophagus (arrow) (HEX40).**

## REFERENCES


[1] Ergun S, Tewfik T, Daniel S. Tracheal agenesis: a rare but fatal congenital anomaly. McGill J Med MJM Int Forum Adv Med Sci Stud 2011;13(1):10.

[2] De Groot-van der Mooren MD, Haak MC, Lakeman P, Cohen-Overbeek TE, van der Voorn JP, Bretschneider JH, et al. Tracheal agenesis: approach towards this severe diagnosis. Case report and review of the literature. Eur J Pediatr Mar 2012;171(3):425–31.

[3] Smith MM, Huang A, Labbé M, Lubov J, Nguyen LHP. Clinical presentation and airway management of tracheal atresia: a systematic review. Int J Pediatr Otorhinolaryngol 2017;101:57–64.

[4] Hedrick MH, Ferro MM, Filly RA, Flake AW, Harrison MR, Adzick NS. Congenital high airway obstruction syndrome (CHAOS): a potential for perinatal intervention. J Pediatr Surg 1994;29(2):271–4.

[5] Lim F-Y, Crombleholme TM, Hedrick HL, Flake AW, Johnson MP, Howell LJ, et al. Congenital high airway obstruction syndrome: natural history and management. J Pediatr Surg 2003;38(6):940–5.

[6] Sattler C, Chiao F, Stein D, Murphy D. Life-saving esophageal intubation in neonate with undiagnosed tracheal agenesis: a case report. Case Rep 2017;9(1):31–4.

[7] Floyd J, Campbell DC, Dominy DE. Agenesis of the trachea. Am Rev Respir Dis Oct 1962;86:557–60.

[8] Faro RS, Goodwin CD, Organ CH, Hall RT, Holder TM, Ashcraft KW, et al. Tracheal Agenesis. Ann Thorac Surg 1979;28(3):295–9.

[9] Park B-J, Kim MS, Yang J-H, Jun T-G. Tracheal agenesis reconstruction with external esophageal stenting: postoperative results and complications. Korean J Thorac Cardiovasc Surg 2015;48(6):439–42.

[10] Das BB, Nagaraj A, Rao AH, Rajegowda BK. Tracheal agenesis: report of three cases and review of the literature. Am J Perinatol 2002;19(7):395–400.